

### **Supplemental Data**

**Supplementary Table E1.** Contributors. The USIDNET query provided information for 199 total patients with a diagnosis of CGD. Information was provided by 27 providers from 26 enrolling centers across the United States.

<b>Institution Name</b>	<b>N</b>	<b>%</b>
NIH/NHGRI/NIAID	113	56.8%
The Children's Hospital of Philadelphia	17	8.5%
Seattle Children's Hospital	7	3.5%
Mount Sinai School of Medicine	6	3.0%
USIDNET/Lurie Children's Hospital of Chicago	6	3.0%
Children's Hospital of Michigan/Wayne State Univ.	6	3.0%
Cincinnati Children's Hospital Medical Center	6	3.0%
University of Utah/Primary Children's Hospital	5	2.5%
University of California, San Francisco	4	2.0%
Advocate Children's Hospital	4	2.0%
University of Iowa	3	1.5%
Duke University Medical Center	2	1.0%
University of California, San Francisco	2	1.0%
Levine Children's Hospital	2	1.0%
University of Miami	2	1.0%
University of California, San Francisco	2	1.0%
University of California, San Francisco	1	0.5%
Nationwide Children's Hospital	1	0.5%
Emory University/Children's Healthcare of Atlanta	1	0.5%
Mayo Clinic	1	0.5%
Children's Hospital of Orange County	1	0.5%
Joe DiMaggio Children's Hospital/Memorial Healthcare	1	0.5%
Massachusetts General and USF	1	0.5%
Children's National	1	0.5%
Washington University/St. Louis Children's Hospital	1	0.5%
Allergy Partners of North Texas	1	0.5%
Children's Hospital of Minnesota	1	0.5%
University of South Florida/All Children's Hospital	1	0.5%

**Total** **199** **100%**

**Supplementary Table E2.** Number of patients with missing demographic information (out of 194 total patients).

	IBD (n=96 patients)	Non-IBD (n=98 patients)
<b>DOB</b>	0	0
<b>Living or Dead</b>	2	3
<b>Age of Death</b>	1	0
<b>Onset of Symptoms</b>	23	30
<b>Age diagnosed</b>	18	33
<b>Genetic mutation</b>	12	12
<b>Unknown ethnicity</b>	9	12

**Supplementary Table E3.** Cohort mortality.

	Age of	Ethnicity and	Genetic
Subject ID	Cause of Death	HSCT	IBD
	Death	Sex	Mutation

10004	2.5	Hispanic/Latino Male	Unknown	Infection	No	Yes
8246	4.8	White/Caucasian Male	CYBB (gp91-phox)	Acute GVHD/multiple organ failure	Yes	Yes
3819	6.2	White/Caucasian Male	CYBB (gp91-phox)	- Aplasia - Disseminated HHV6 with limbic encephalitis	Yes	No
10331	10.3	Unknown Male	Unknown	Unknown	No	No
3464	15.8	White/Caucasian Male	Unknown	Complication from HSCT	Yes	Yes
2322	17	Unknown Male	CYBB (gp91-phox)	Car accident	No	No
9058	19.3	White/Caucasian Male	CYBB (gp91-phox)	Progressive pulmonary infiltrates and hyperemic respiratory failure	Yes	Yes
10334	19.7	Unknown Male	CYBA (p22phox)	Arrest during pneumonectomy with massive hemorrhage	No	No
1431	23.9	White/Caucasian Male	CYBB (gp91-phox)	<i>Paecilomyces</i> pneumonia	No	No
6971	23.9	White/Caucasian Male	CYBA (p22phox)	- Post-HSCT GVHD - Multiple infections: CMV, <i>C. diff</i> , BK cystitis, <i>Legionella</i> peritonitis, <i>Candida glabrata</i> fungemia, VRE bacteremia, end stage nephropathy, refractory thrombocytopenia, respiratory failure	Yes	No
3762	24.5	White/Caucasian Male	CYBB (gp91-phox)	- Hepatocellular carcinoma - Hemorrhage	No	Yes
3892	26.1	Black/African American Male	CYBB (gp91-phox)	-Sepsis	No	No

10900	31.6	Unknown Male	CYBB (gp91-phox)	-Infection: <i>Burkholderia</i> pneumonia, <i>Phellinus</i> cellulitis -Engraftment syndrome -Refractory hypoxemia -Renal and hepatic failure	Yes	No
10333	33	Unknown Male	CYBB (gp91-phox)	-Uremia, electrolyte abnormalities	Yes	Yes
7652	37.4	Black/African American Male	CYBB (gp91-phox)	-Hypoxic respiratory failure s/p BMT -Acute on chronic renal failure - <i>Nocardia</i> pneumonia	Yes	No
6977	48.1	White/Caucasian Male	CYBA (p22phox)	-Malignancy -AV fistula and arterial clot	No	Yes
1430	Unknown (born in 1984)	White/Caucasian Female	CYBA (p22phox)	-Hemorrhage secondary to fungal infection	No	Yes

**Supplementary Table E4.** Patient genotype.

Gene	Overall (n=194)	IBD (n=96)	Non-IBD (n=98)
<b><i>CYBB</i> gp91phox</b>	63.4%	67.7%	59.2%
Male	121	63	58
Female	2	2	0
<b><i>NCF1</i> p47phox</b>	13.9%	10.4%	17.3%
Male	18	8	10
Female	9	2	7
<b><i>CYBA</i> p22phox</b>	7.7%	6.3%	9.2%
Male	10	4	6
Female	5	2	3
<b><i>NCF2</i> p67phox</b>	2.6%	3.1%	2.0%
Male	2	0	2
Female	3	3	0
<b><i>NCF4</i></b>	0%	0%	0%
Male	0	0	0
Female	0	0	0
<b>Unknown</b>	12.4%	12.5% 11	12.2%
Male	18	1	7
Female	6		5

**Supplementary Table E5.** Other infections.

**Supplementary Table E6.** Additional prophylaxis\*.

	IBD	Non-
<b>Anti-infective</b>		
	IBD	
<b>Acyclovir</b>	10	8
<b>Pentamidine</b>	3	4
<b>Meropenem</b>	0	4
<b>Levofloxacin</b>	7	3
<b>Isavuconazonium</b>	5	3
<b>Ciprofloxacin</b>	3	3
<b>Azithromycin</b>	2	2

<b>Vancomycin</b>	2	2
<b>Caspofungin</b>	0	2
<b>Amphotericin</b>	1	2
<b>Doxycycline</b>	1	3
<b>Fluconazole</b>	4	1
<b>Clindamycin</b>	3	1
<b>Cefdinir</b>	2	1
<b>Daptomycin</b>	0	1
<b>Amoxicillin clavulanate</b>	2	2
<b>Cefepime</b>	0	1
<b>Ceftriaxone</b>	0	1
<b>Cephalexin</b>	1	1
<b>Ciclopirox</b>	0	1
<b>Terbinafine</b>	1	1
<b>Micafungin</b>	2	1
<b>Dicloxacillin</b>	0	1
<b>Amoxicillin</b>	2	0
<b>Atovaquone</b>	1	0
<b>Clarithromycin</b>	1	0
<b>Rifaximin</b>	1	0
<b>Tedizolid</b>	1	0
<b>Posaconazole</b>	1	0

\*Drugs listed above were categorized as prophylaxis by the submitting USIDNET center and could be administered as “continuous,” “intermittent,” or “rotating” prophylaxis.

**Supplementary Table E7.** Immunomodulator drug therapies

Category	# Patients by Category (% of total CGD cohort) (n=194)	Drug	# Patients (% of each group)		
			Total CGD Cohort (n=194)	IBD (n=96)	Non-IBD (n=98)
<b>IFN-gamma</b>	80 (41.2%)	IFN-gamma	80 (41.2%)	40 (41.7%)	40 (40.8%)
<b>Corticosteroids</b>	80 (41.2%)	Corticosteroids	80 (41.2%)	60 (62.5%)	20 (20.4%)
<b>Other</b>	51 (26.3%)	Sirolimus	20 (10.3%)	12 (12.5%)	8 (8.2%)
		Mesalamine	18 (9.3%)	18 (18.8%)	0
		Tacrolimus	9 (4.6%)	6 (6.3%)	4 (4.1%)
		Sulfasalazine	1 (0.5%)	1 (1.0%)	0

		Balsalazide	1 (0.5%)	1 (1.0%)	0
		Cyclosporine	1 (0.5%)	1 (1.0%)	0
		Thalidomide	1 (0.5%)	1 (1.0%)	0
Antimetabolites/DMARDs	29 (14.9%)	Azathioprine	9 (4.6%)	9 (9.4%)	0
		6-Mercaptopurine	8 (4.1%)	7 (7.3%)	1 (1.0%)
		Mycophenolate	5 (2.6%)	4 (4.2%)	1 (1.0%)
		Hydroxychloroquine	4 (2.1%)	2 (2.1%)	2 (2.0%)
		Methotrexate	3 (1.5%)	2 (2.1%)	1 (1.0%)
Other monoclonal antibodies	14 (7.2%)	Vedolizumab	11 (5.7%)	11 (11.5%)	0
		Basiliximab	1 (0.5%)	1 (1.0%)	0
		Rituximab	5 (2.6%)	4 (4.2%)	1 (1.0%)
		Eculizumab	1 (0.5%)	0	1 (1.0%)
Anti-cytokine monoclonal antibodies	14 (7.2%)	TNF-alpha inhibitor	8 (4.1%)	7 (7.3%)	1 (1.0%)
		Anakinra	4 (2.1%)	3 (3.1%)	1 (1.0%)
		Ustekinumab	2 (1.0%)	2 (2.1%)	0
Small molecule/JAK inhibitor	5 (2.6%)	Ruxolitinib	1 (0.5%)	1 (1.0%)	0
		<b>Total Occurrences</b>	<b>273</b>	<b>193</b>	<b>81</b>